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# SWISS-PROT: O60214

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ID TRT2\_HUMAN STANDARD; PRT; 297 AA.  
AC P45379; Q99596; Q99597; O60214;  
DT 01-NOV-1995 (Rel. 32, Created)  
DT 15-JUN-2002 (Rel. 41, Last sequence update)  
DT 15-JUN-2002 (Rel. 41, Last annotation update)  
DE Troponin T, cardiac muscle isoforms (TnTC).  
GN TNNT2.  
OS Homo sapiens (Human).  
OC Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;  
OC Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.  
OX NCBI\_TaxID=9606;  
RN [1]  
RP SEQUENCE FROM N.A. (ISOFORM 6).  
RC TISSUE=Heart;  
RX MEDLINE=93345675; PubMed=8344420; [NCBI, ExPASy, EBI, Israel, Japan]  
RA Mesnard L., Samson F., Espinasse I., Durand J., Neveux J.-Y.,  
RA Mercadier J.-J.;  
RT "Molecular cloning and developmental expression of human cardiac  
RT troponin T.";  
RL FEBS Lett. 328:139-144(1993).  
RN [2]  
RP SEQUENCE FROM N.A. (ISOFORMS 1 AND 6).  
RC TISSUE=Heart muscle;  
RX MEDLINE=94375053; PubMed=8088824; [NCBI, ExPASy, EBI, Israel, Japan]  
RA Townsend P.J., Farza H., Macgeoch C., Spurr N.K., Wade R.,  
RA Gahlman R., Yacoub M.H., Barton P.J.R.;  
RT "Human cardiac troponin T: identification of fetal isoforms and  
RT assignment of the TNNT2 locus to chromosome 1q.";  
RL Genomics 21:311-316(1994).  
RN [3]  
RP SEQUENCE FROM N.A. (SPLICED ISOFORMS).  
RC TISSUE=Fetal heart;  
RX MEDLINE=96129582; PubMed=8576938; [NCBI, ExPASy, EBI, Israel, Japan]  
RA Townsend P.J., Barton P.J.R., Yacoub M.H., Farza H.;  
RT "Molecular cloning of human cardiac troponin T isoforms: expression in  
RT developing and failing heart.";  
RL J. Mol. Cell. Cardiol. 27:2223-2236(1995).  
RN [4]  
RP SEQUENCE FROM N.A. (ISOFORMS 1; 6; 7 AND 8).  
RC TISSUE=Heart;  
RX MEDLINE=95202803; PubMed=7534662; [NCBI, ExPASy, EBI, Israel, Japan]  
RA Anderson P.A., Greig A., Mark T.M., Malouf N.N., Oakeley A.E.,  
RA Ungerleider R.M., Allen P.D., Kay B.K.;  
RT "Molecular basis of human cardiac troponin T isoforms expressed in  
RT the developing, adult, and failing heart.";  
RL Circ. Res. 76:681-686(1995).  
RN [5]

RP SEQUENCE FROM N.A. (ISOFORMS 1; 2; 3; 4; 5; 6 AND 10).  
 RC TISSUE=Fetal heart;  
 RX MEDLINE=95202804; PubMed=7895342; [NCBI, ExpASY, EBI, Israel, Japan]  
 RA Mesnard L., Logeart D., Taviaux S., Diriong S., Mercadier J.-J.,  
 RA Samson F.;  
 RT "Human cardiac troponin T: cloning and expression of new isoforms in  
 RT the normal and failing heart."  
 RL Circ. Res. 76:687-692(1995).  
 RN [6]  
 RP SEQUENCE FROM N.A. (ISOFORM 6), AND VARIANT FHC ILE-119.  
 RC TISSUE=Heart muscle;  
 RX MEDLINE=98141687; PubMed=9482583; [NCBI, ExpASY, EBI, Israel, Japan]  
 RA Gerull B., Osterziel K.-J., Witt C., Dietz R., Thierfelder L.;  
 RT "A rapid protocol for cardiac troponin T gene mutation detection in  
 RT familial hypertrophic cardiomyopathy."  
 RL Hum. Mutat. 11:179-182(1998).  
 RN [7]  
 RP SEQUENCE FROM N.A. (ISOFORM 6).  
 RA D'Cruz L.G., Oberoi J., Mughal F., Steffensen U., Steffensen M.,  
 RA Kubo T., Mogensen J., McKoy G., O'Donnoghue A., Pondel M.,  
 RA McKenna W.J., Carter N.D., Baboonian C.;  
 RT "Genomic organization of the human cardiac troponin T gene (TNNT2) and  
 RT characterization of the candidate promoter region."  
 RL Submitted (JUN-2001) to the EMBL/GenBank/DDBJ databases.  
 RN [8]  
 RP SEQUENCE OF 190-228 AND 230-287 FROM N.A.  
 RC TISSUE=Blood;  
 RA Farza H., Townsend P.J.;  
 RL Submitted (FEB-1997) to the EMBL/GenBank/DDBJ databases.  
 RN [9]  
 RP SEQUENCE OF 69-75 AND 176-181.  
 RC TISSUE=Heart;  
 RX MEDLINE=96007936; PubMed=7498159; [NCBI, ExpASY, EBI, Israel, Japan]  
 RA Kovalyov L.I., Shishkin S.S., Efimochkin A.S., Kovalyova M.A.,  
 RA Ershova E.S., Egorov T.A., Musalyamov A.K.;  
 RT "The major protein expression profile and two-dimensional protein  
 RT database of human heart."  
 RL Electrophoresis 16:1160-1169(1995).  
 RN [10]  
 RP VARIANTS FHC ASN-88 AND GLN-101.  
 RX MEDLINE=94265260; PubMed=8205619; [NCBI, ExpASY, EBI, Israel, Japan]  
 RA Thierfelder L., Watkins H., Macrae C., Lamas R., McKenna W.,  
 RA Vosberg H.-P., Seidman J.G., Seidman C.E.;  
 RT "Alpha-tropomyosin and cardiac troponin T mutations cause familial  
 RT hypertrophic cardiomyopathy: a disease of the sarcomere."  
 RL Cell 77:701-712(1994).  
 RN [11]  
 RP VARIANTS FHC.  
 RX MEDLINE=95206332; PubMed=7898523; [NCBI, ExpASY, EBI, Israel, Japan]  
 RA Watkins H., McKenna W.J., Thierfelder L., Suk H.J., Anan R.,  
 RA O'Donoghue A., Spirito P., Matsumori A., Moravec C.S., Seidman J.G.,  
 RA Seidman C.E.;  
 RT "Mutations in the genes for cardiac troponin T and alpha-tropomyosin  
 RT in hypertrophic cardiomyopathy."  
 RL New Engl. J. Med. 332:1058-1064(1995).  
 RN [12]  
 RP VARIANT FHC PRO-287.  
 RA Erdmann J., Wischke S., Kallisch H., Riedel K., Heidenreich M.,  
 RA Fleck E., Regitz-Zagrosek V.;  
 RT "A novel missense Arg 278 Pro mutation in the troponin T gene

RT (TNNT2).";  
 RL Hum. Mutat. 12:364-364(1998).  
 RN [13]  
 RP VARIANT FHC LEU-103.  
 RX MEDLINE=99457222; PubMed=10525521; [NCBI, ExpASY, EBI, Israel, Japan]  
 RA Varnava A., Baboonian C., Davison F., de Cruz L., Elliott P.M.,  
 RA Davies M.J., McKenna W.J.;  
 RT "A new mutation of the cardiac troponin T gene causing familial  
 RT hypertrophic cardiomyopathy without left ventricular hypertrophy."  
 RL Heart 82:621-624(1999).

CC -!- FUNCTION: TROPONIN T IS THE TROPOMYOSIN-BINDING SUBUNIT OF  
 CC TROPONIN, THE THIN FILAMENT REGULATORY COMPLEX WHICH CONFERS  
 CC CALCIUM-SENSITIVITY TO STRIATED MUSCLE ACTOMYOSIN ATPASE ACTIVITY.  
 CC -!- ALTERNATIVE PRODUCTS: At least 10 isoforms; isoform 1/TNT1 (shown  
 CC here), 2, 3, 4, 5, 6/TNT3, 7/TNT4, 8/TNT2, 9 and 10; may be  
 CC produced by alternative splicing.  
 CC -!- TISSUE SPECIFICITY: Heart. The fetal heart shows a greater  
 CC expression in the atrium than in the ventricle, while the adult  
 CC heart shows a greater expression in the ventricle than in the  
 CC atrium. Isoform 6 predominates in normal adult heart. Isoforms 1,  
 CC 7 and 8 are expressed in fetal heart. Isoform 7 is also expressed  
 CC in failing adult heart.  
 CC -!- DISEASE: DEFECTS IN TNNT2 ARE ONE OF THE CAUSES OF FAMILIAL  
 CC HYPERTROPHIC CARDIOMYOPATHY (FHC) WHICH IS AN AUTOSOMAL DOMINANT  
 CC DISORDER CHARACTERIZED BY INCREASED MYOCARDIAL MASS WITH MYOCYTE  
 CC AND MYOFIBRILLAR DISARRAY. THIS FORM OF FHC IS KNOWN AS CMH2.  
 CC IT IS A DISEASE OF THE SARCOMERE.  
 CC -!- SIMILARITY: BELONGS TO THE TROPONIN T FAMILY.

CC -----  
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 CC -----

DR EMBL; S64668; AAB27731.1; ALT\_SEQ. [EMBL / GenBank / DDBJ] [CoDingSequence]  
 DR EMBL; X74819; CAA52818.1; -. [EMBL / GenBank / DDBJ] [CoDingSequence]  
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DR EMBL; AF004421; AAC39590.1; JOINED. [EMBL / GenBank / DDBJ] [CoDingSequence]  
 DR EMBL; AY044273; AAK92231.1; -. [EMBL / GenBank / DDBJ] [CoDingSequence]  
 DR EMBL; S71128; AAB30957.1; -. [EMBL / GenBank / DDBJ] [CoDingSequence]  
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 DR Genew; HGNC:11949; TNNT2.  
 DR MIM; 191045; -. [NCBI / EBI]  
 DR GeneCards; TNNT2.  
 DR GeneLynx; TNNT2.  
 DR Ensembl; P45379. [Entry / Contig view]  
 DR SOURCE; TNNT2.  
 DR MIM; 115195; -. [NCBI / EBI]  
 DR InterPro; IPR001978; Troponin.  
 DR InterPro; Graphical view of domain structure.  
 DR Pfam; PF00992; Troponin; 1.  
 DR ProDom [Domain structure / List of seq. sharing at least 1 domain]  
 DR BLOCKS; O60214.  
 DR ProtoNet; O60214.  
 DR ProtoMap; O60214.  
 DR PRESAGE; P45379.  
 DR DIP; P45379.  
 DR ModBase; P45379.  
 DR SWISS-2DPAGE; GET REGION ON 2D PAGE.  
 KW Muscle protein; Alternative splicing; Multigene family;  
 KW Phosphorylation; Disease mutation; Polymorphism; Cardiomyopathy.  
 FT INIT MET 0 0  
 FT MOD\_RES 1 1 PHOSPHORYLATION (BY CK2) (BY SIMILARITY).  
 FT VARSPLIC 17 21 MISSING (IN ISOFORM 8).  
 FT VARSPLIC 17 31 MISSING (IN ISOFORM 7).  
 FT VARSPLIC 22 31 MISSING (IN ISOFORM 6).  
 FT VARSPLIC 22 22 MISSING (IN ISOFORM 2 AND ISOFORM 4).  
 FT VARSPLIC 53 53 MISSING (IN ISOFORM 3 AND ISOFORM 4).  
 FT VARSPLIC 98 136 MISSING (IN ISOFORM 9).  
 FT VARSPLIC 200 200 MISSING (IN ISOFORM 5).  
 FT VARSPLIC 200 202 MISSING (IN ISOFORM 10).  
 FT VARIANT 88 88 I -> N (IN FHC CMH2).  
 FT VARIANT 101 101 /FTId=VAR\_007605.  
 FT VARIANT 103 103 R -> Q (IN FHC CMH2).  
 FT VARIANT 119 119 /FTId=VAR\_007606.  
 FT VARIANT 138 138 R -> L (IN FHC CMH2).  
 FT VARIANT 169 169 /FTId=VAR\_009194.  
 FT VARIANT 172 172 F -> I (IN FHC CMH2).  
 FT VARIANT 248 248 /FTId=VAR\_007607.  
 FT VARIANT 253 253 R -> K.  
 FT VARIANT 262 262 /FTId=VAR\_013021.  
 FT VARIANT 287 287 MISSING (IN FHC CMH2).  
 FT VARIANT 287 287 /FTId=VAR\_007608.  
 FT VARIANT 287 287 E -> K (IN FHC CMH2).  
 FT VARIANT 287 287 /FTId=VAR\_007609.  
 FT VARIANT 287 287 S -> T.  
 FT VARIANT 287 287 /FTId=VAR\_013022.  
 FT VARIANT 287 287 E -> D (IN FHC CMH2).  
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 FT VARIANT 287 287 K -> R.  
 FT VARIANT 287 287 /FTId=VAR\_007611.  
 FT VARIANT 287 287 R -> C (IN FHC CMH2).  
 FT VARIANT 287 287 /FTId=VAR\_007612.  
 FT VARIANT 287 287 R -> P (IN FHC CMH2).  
 FT VARIANT 287 287 /FTId=VAR\_007613.  
 FT CONFLICT 241 241 K -> E (IN REF. 8).  
 FT CONFLICT 262 262 K -> R (IN REF. 8).

SQ SEQUENCE 297 AA; 35792 MW; 66FDCD1EE4A3C965 CRC64;  
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AKEAEDGPME ESKPKPRSFM PNLVPPKIPD GERVDFDDIH RKRMEKDLNE LQALIEAHFE  
NRKKEEEELV SLKDRIERRR AERAEQQRIR NEREKERQNR LAEERARREE EENRRKAEDE  
ARKKKALSNM MHFGGYIQKQ AQTERKSGKR QTEREKKKKI LAERRKVLAI DHLNEDQLRE  
KAKELWQSIY NLEAEKFDLQ EKFKQQKYEI NVLRNRINDN QKVSCTRGA KVTGRWK

//

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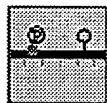
Direct BLAST submission at [NCBI \(Bethesda, USA\)](#)



[ScanProsite](#), [MotifScan](#)



Tools Sequence analysis tools: [ProtParam](#), [ProtScale](#),  
[Compute pI/Mw](#), [PeptideMass](#), [PeptideCutter](#),  
[Dotlet \(Java\)](#)







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<b>ProDom Release 2001.3</b>  <b>Domain PD015088</b>	 Graphic representation of all proteins containing this domain.
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	<a href="#">MSF Alignment in MSF format.</a>
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Run Predict Protein server with this domain 	


Most frequent protein names	TRT2(7)
Commentary (automatic)	MUSCLE TROPONIN CARDIAC SKELETAL PHOSPHORYLATION ISOFORMS SPLICING ALTERNATIVE FAMILY
Alignment length	72
Number of domains in family	23
Consistency indicator	DIAMETER: 124 PAM RADIUS OF GYRATION: 38 PAM SEQUENCE CLOSEST TO CONSENSUS: TRT2_BOVIN 58-107 (distance: 16 PAM)

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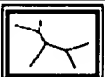
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Other descriptions of the family  
corresponding to this alignment



Sequence ID	start	end	weight	10	20	30	40	50
1 O13096_SALTR	42	104	4.27	.....	EETPAEEASGETQDSKAKP-KSFMPNVAPPKLPEGDGKVD	FDDLHR		
5 TTRT1_HUMAN	.	.	2.27	.....	REERPKPSRPVVPPLIPPKEGE-RVDFDDIHR			
4 TTRT2_RAT	.	.	2.01	VEEVGPDEEAKDAEEGPVEDTKPKPSRLFMPNLVPPKIPDGE-RVDFDDIHR				
8 TTRT2_HUMAN	.	.	4.25	.....	EEGDREQEPGPGGEESKPKPPRPFMPNLVPPKIPDGE-RVDFDDIHR			
2 O93376_SALSA	.	.	3.03	.....	EQHFEEEEKPK----	FKPTAKAPKIPDGE-KVDFDDIQK		
1 Q99L89_MOUSE	60	93	0.30	.....	PKIPEGE-KVDFDDIQK			
1 Q9QZ47_MOUSE	59	92	0.45	.....	PKIPEGE-KVDFDDIQK			
1 Q25148_HALRO	3	63	6.42	.....	GEEEEPQQHHEDVEKP-MPRHSTTSIPRLPDGE-KVDLDVITQ			
23 Consensus			23.00	VEEVGPDEETGEEEPGQHHEDKPKPSRPFMPNLAPPKIPDGE	GKVD	FDDIHR		

Minimal distance between sequences (in PAM)	<input type="text" value="20"/>
Maximal number of clusters	<input type="text" value="12"/>
If possible, clusterIDs should contain the following string (e.g.: human)	<input type="text"/>

To display a new alignment with these parameters,  
or click [here](#) to display all sequences in the family

[Submit Query](#)

Eukaryotic subfamily root	Bacterial subfamily root	Archaeal subfamily root	Viral subfamily root	Root of a subfamily that goes across domains of life
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


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<b>ProDom Release 2001.3</b>  <b>Domain PD349636</b>	 Graphic representation of all proteins containing this domain.
	 Display the family as a tree.
	<a href="#">MSF Alignment in MSF format.</a>
	Build an ESPript view 
	Run Predict Protein server with this domain 

Most frequent protein names	TRT2(6) TRT3(5)
Commentary (automatic)	MUSCLE TROPONIN SKELETAL PHOSPHORYLATION FAST MULTIGENE FAMILY ISOFORMS SPLICING
Alignment length	32
Number of domains in family	32
Consistency indicator	DIAMETER: 35 PAM RADIUS OF GYRATION: 14 PAM SEQUENCE CLOSEST TO CONSENSUS: Q9QUP7_MOUSE 88-117 (distance: 7 PAM)


### InterPro

ID	Accession number
"Troponin"	IPR001978

### Pfam-A

ID	Accession number
Troponin	PF00992

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Other descriptions of the family  
corresponding to this alignment



Sequence ID	start	end	weight	10	20	30
2 <a href="#">TRT2_CHICK</a>	.	.	7.39	EHRKKEEEELISLKDRIEQRRRAERAEQQRIRS		
10 <a href="#">TRT2_HUMAN</a>	.	.	6.60	ENRKKEEEELVSLKDRIEKRRRAERAEQQRIRN		
14 <a href="#">TRT3_RAT</a>	.	.	5.54	EARKKEEEELVALKERIEKRRRAERAEQQRIRA		
1 <a href="#">Q9I8U9_BRARE</a>	57	88	5.00	EHRQKEEEELIALRERIEKRRSERAEQQRIRT		
5 <a href="#">TRT1_HUMAN</a>	.	.	7.45	. . RKKEEEELVALKDRIERRRSEAEQQRFRT		
32 Consensus			31.98	EHRKKEEEELIALKDRIEKRRRAERAEQQRIRT		

Minimal distance between sequences (in PAM)	<input type="text" value="20"/>
Maximal number of clusters	<input type="text" value="12"/>
If possible, clusterIDs should contain the following string (e.g.: human)	<input type="text"/>

To display a new alignment with these parameters,  
or click [here](#) to display all sequences in the family

[Submit Query](#)

<b>Eukaryotic subfamily root</b>	<b>Bacterial subfamily root</b>	<b>Archaeal subfamily root</b>	<b>Viral subfamily root</b>	<b>Root of a subfamily that goes across domains of life</b>
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## General information about the entry

Entry name **TRT2\_HUMAN**  
 Primary accession number **P45379**  
 Secondary accession numbers Q99596 Q99597 O60214  
 Entered in SWISS-PROT in Release 32, November 1995  
 Sequence was last modified in Release 41, June 2002  
 Annotations were last modified in Release 41, June 2002

## Name and origin of the protein

Protein name **Troponin T, cardiac muscle isoforms**  
 Synonym **TnTC**  
 Gene name **TNNT2**  
 From **Homo sapiens (Human) [TaxID: 9606]**  
 Taxonomy **Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.**

## References

- [1] SEQUENCE FROM NUCLEIC ACID (ISOFORM 6).  
**TISSUE=Heart;**  
 MEDLINE=93345675; PubMed=8344420; [NCBI, ExPASy, EBI, Israel, Japan]  
Mesnard L., Samson F., Espinasse I., Durand J., Neveux J.-Y., Mercadier J.-J.;  
 "Molecular cloning and developmental expression of human cardiac troponin T."; *FEBS Lett.* 328:139-144(1993).
- [2] SEQUENCE FROM NUCLEIC ACID (ISOFORMS 1 AND 6).  
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 "Human cardiac troponin T: identification of fetal isoforms and assignment of the TNNT2 locus to chromosome 1q."; *Genomics* 21:311-316(1994).
- [3] SEQUENCE FROM NUCLEIC ACID (SPLICED ISOFORMS).  
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TISSUE=Blood;

Farza H., Townsend P.J.;

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TISSUE=Heart;

MEDLINE=96007936; PubMed=7498159; [NCBI, ExPASy, EBI, Israel, Japan]

Kovalyov L.I., Shishkin S.S., Efimochkin A.S., Kovalyova M.A., Ershova E.S., Egorov T.A., Musalyamov A.K.;

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Cell 77:701-712(1994).

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MEDLINE=95206332; PubMed=7898523; [NCBI, ExPASy, EBI, Israel, Japan]  
Watkins H., McKenna W.J., Thierfelder L., Suk H.J., Anan R., O'Donoghue A., Spirito P.,  
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 "Mutations in the genes for cardiac troponin T and alpha-tropomyosin in hypertrophic  
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 "A novel missense Arg 278 Pro mutation in the troponin T gene (TNNT2).";  
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 without left ventricular hypertrophy.";  
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## Comments

- **FUNCTION:** TROPONIN T IS THE TROPOMYOSIN-BINDING SUBUNIT OF TROPONIN, THE THIN FILAMENT REGULATORY COMPLEX WHICH CONFERS CALCIUM-SENSITIVITY TO STRIATED MUSCLE ACTOMYOSIN ATPASE ACTIVITY.
- **ALTERNATIVE PRODUCTS:** At least 10 isoforms; isoform 1/TNT1 (shown here), 2, 3, 4, 5, 6/TNT3, 7/TNT4, 8/TNT2, 9 and 10; may be produced by alternative splicing.
- **TISSUE SPECIFICITY:** Heart. The fetal heart shows a greater expression in the atrium than in the ventricle, while the adult heart shows a greater expression in the ventricle than in the atrium. Isoform 6 predominates in normal adult heart. Isoforms 1, 7 and 8 are expressed in fetal heart. Isoform 7 is also expressed in failing adult heart.
- **DISEASE:** DEFECTS IN TNNT2 ARE ONE OF THE CAUSES OF FAMILIAL HYPERTROPHIC CARDIOMYOPATHY (FHC) WHICH IS AN AUTOSOMAL DOMINANT DISORDER CHARACTERIZED BY INCREASED MYOCARDIAL MASS WITH MYOCYTE AND MYOFIBRILLAR DISARRAY. THIS FORM OF FHC IS KNOWN AS CMH2. IT IS A DISEASE OF THE SARCOMERE.
- **SIMILARITY:** BELONGS TO THE TROPONIN T FAMILY.

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## Cross-references

S64668; AAB27731.1; ALT_SEQ.	[EMBL / GenBank / DDBJ] [CoDingSequence]
X74819; CAA52818.1; -.	[EMBL / GenBank / DDBJ] [CoDingSequence]
L40162; AAA67422.1; -.	[EMBL / GenBank / DDBJ] [CoDingSequence]
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 MIM [191045 \[NCBI / EBI\].](#)  
[115195 \[NCBI / EBI\].](#)  
 GeneCards [TNNT2.](#)  
 GeneLynx [TNNT2.](#)  
 SOURCE [TNNT2; Homo sapiens.](#)  
 Ensembl [P45379; Homo sapiens. \[Entry / Contig view\]](#)  
 InterPro [IPR001978; Troponin.](#)  
[Graphical view of domain structure.](#)  
 Pfam [PF00992; Troponin; 1.](#)  
 ProDom [\[Domain structure / List of seq. sharing at least 1 domain\].](#)  
 BLOCKS [P45379.](#)  
 ProtoNet [P45379.](#)  
 ProtoMap [P45379.](#)  
 PRESAGE [P45379.](#)  
 DIP [P45379.](#)  
 ModBase [P45379.](#)  
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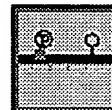
**Keywords**

**Muscle protein; Alternative splicing; Multigene family; Phosphorylation; Disease mutation; Polymorphism; Cardiomyopathy.**

**Features**

Key	From	To	Length	Description
INIT_MET	0	0		
MOD_RES	<u>1</u>	<u>1</u>		PHOSPHORYLATION (BY CK2) (BY SIMILARITY) .
VARSPLIC	<u>17</u>	<u>21</u>		MISSING (IN <u>ISOFORM 8</u> ) .
VARSPLIC	<u>17</u>	<u>31</u>		MISSING (IN <u>ISOFORM 7</u> ) .
VARSPLIC	<u>22</u>	<u>31</u>		MISSING (IN <u>ISOFORM 6</u> ) .
VARSPLIC	<u>22</u>	<u>22</u>		MISSING (IN <u>ISOFORM 2</u> AND <u>ISOFORM 4</u> ) .
VARSPLIC	<u>53</u>	<u>53</u>		MISSING (IN <u>ISOFORM 3</u> AND

VARSP LIC	<u>98</u>	<u>136</u>	ISOFORM 4).
VARSP LIC	<u>200</u>	<u>200</u>	MISSING (IN ISOFORM 9).
VARSP LIC	<u>200</u>	<u>202</u>	MISSING (IN ISOFORM 5).
VARIANT	<u>88</u>	<u>88</u>	MISSING (IN ISOFORM 10).
VARIANT	<u>101</u>	<u>101</u>	I -> N (IN FHC CMH2).
VARIANT	<u>103</u>	<u>103</u>	/FTId=VAR_007605.
VARIANT	<u>119</u>	<u>119</u>	R -> Q (IN FHC CMH2).
VARIANT	<u>138</u>	<u>138</u>	/FTId=VAR_007606.
VARIANT	<u>169</u>	<u>169</u>	R -> L (IN FHC CMH2).
VARIANT	<u>172</u>	<u>172</u>	/FTId=VAR_009194.
VARIANT	<u>248</u>	<u>248</u>	F -> I (IN FHC CMH2).
VARIANT	<u>253</u>	<u>253</u>	/FTId=VAR_007607.
VARIANT	<u>262</u>	<u>262</u>	R -> K.
VARIANT	<u>287</u>	<u>287</u>	/FTId=VAR_013021.
VARIANT	<u>287</u>	<u>287</u>	MISSING (IN FHC CMH2).
CONFLICT	<u>241</u>	<u>241</u>	/FTId=VAR_007608.
CONFLICT	<u>262</u>	<u>262</u>	E -> K (IN FHC CMH2).
			/FTId=VAR_007609.
			S -> T.
			/FTId=VAR_013022.
			E -> D (IN FHC CMH2).
			/FTId=VAR_007610.
			K -> R.
			/FTId=VAR_007611.
			R -> C (IN FHC CMH2).
			/FTId=VAR_007612.
			R -> P (IN FHC CMH2).
			/FTId=VAR_007613.
			K -> E (IN REF. 8).
			K -> R (IN REF. 8).



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### Sequence information

Length: **297** Molecular weight: **35792** CRC64: **66FDCD1EE4A3C965** [This is a checksum on the AA Da sequence]

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70	80	90	100	110	120
AKEAEDGPME	ESKPKPRSF	PNLVPPKIPD	GERVDFDDIH	RKRMEKDLNE	LQALIEAHFE
130	140	150	160	170	180
NRKKEEEELV	SLKDRIERR	AERAEQQRIR	NEREKERQNR	LAEERARREE	EENRRKAEDE
190	200	210	220	230	240
ARKKKALSNM	MHFGGYIQKQ	AQTERKSGKR	QTEREKKKKI	LAERRKVLAI	DHLNEDQLRE
250	260	270	280	290	
KAKELWQSIY	NLEAEKFDLQ	EKFKQQKYEI	NVLNRNRINDN	QKVSCTRGA	KVTGRWK

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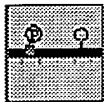
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[ScanProsite](#), [MotifScan](#)



Tools Sequence analysis tools: [ProtParam](#), [ProtScale](#),  
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